# Successful surgical intervention for giant thoracic aortic aneurysm in cutis laxa aortopathy



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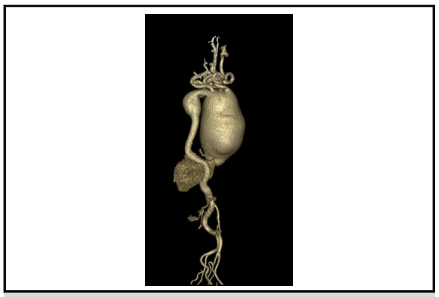


Cutis laxa arterial tortuosity with giant aortic aneurysm from root to descending aorta.

### CENTRAL MESSAGE

We report a giant aneurysm of the thoracic aorta in a child with cutis laxa aortopathy successfully treated with aortic valve repair and thoracic aortic replacement using a custom-made Dacron graft.

Video clip is available online.

Prophylactic aortic surgery remains the most effective method of preventing premature cardiovascular death due to rupture or dissection from hereditary aortopathies.[1,2] Autosomal-recessive cutis laxa type 1B (ARCL 1B) is an extremely rare genetic disorder with life-threatening progressive aortic aneurysmal disease.[3-6] There are no clinical practice guidelines and limited data on surgical intervention in aneurysms extending beyond the ascending aorta in children with cutis laxa, especially with additional aortic leaflet pathology.[2,3]

We report giant aneurysm of the thoracic aorta in a child with cutis laxa aortopathy that was successfully treated with aortic valve repair and thoracic aortic replacement. Informed written consent was taken from the parents for this publication. This case report was exempt from institutional review board approval.

## CASE REPORT

A 7-year-old previously healthy girl (height 117 cm, weight 16 kg), was referred to our unit with aneurysmal dilation of thoracic aorta and severe aortic regurgitation (AR), as demonstrated on transthoracic echocardiogram. Three-dimensional transesophageal echocardiography demonstrated a trileaflet aortic valve with dilated annulus (23 mm, z score +5.34) and a central coaptation defect. The noncoronary cusp was mildly thickened.

Computed tomographic angiography (CTA) demonstrated extensive aortic dilation from aortic root to the arch (maximum diameter 71 mm). This was followed abruptly by a relatively narrow isthmus and a dilated aneurysmal proximal descending aorta (length 50 mm × width 30 mm). Increased tortuosity of the arch vessels and abdominal aorta was noted (Figure 1). Cerebral CTA showed tortuous vessels but no aneurysms or stenosis. Homozygous missense mutation (c. 608A>C; pAsp203Ala) in exon-7 of FBLN4 gene confirmed ARCL IB.

After median sternotomy, cardiopulmonary bypass was instituted with a 7-mm Dacron graft sutured to the right common carotid artery and venous drainage through the right atrium. At 25 °C, the ascending aorta was opened transversely and ostia cardioplegia administered (Video 1). Systematic assessment of leaflet quality and mobility corresponded with transesophageal echocardiography findings. An unmodified 22-mm Dacron

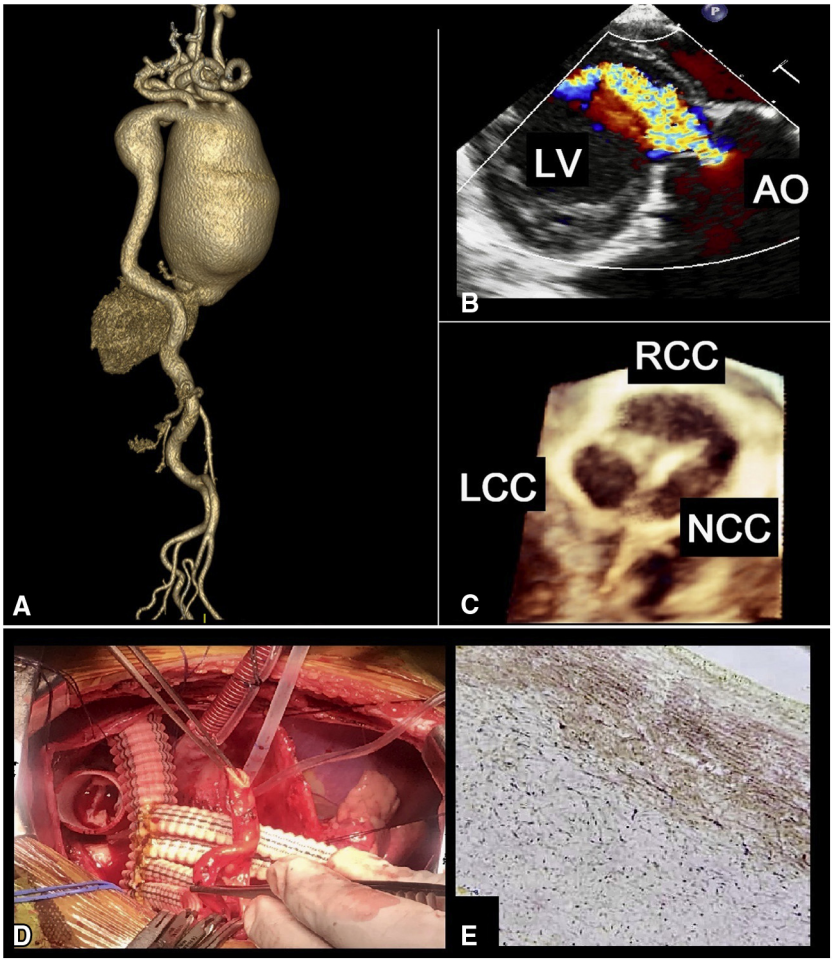
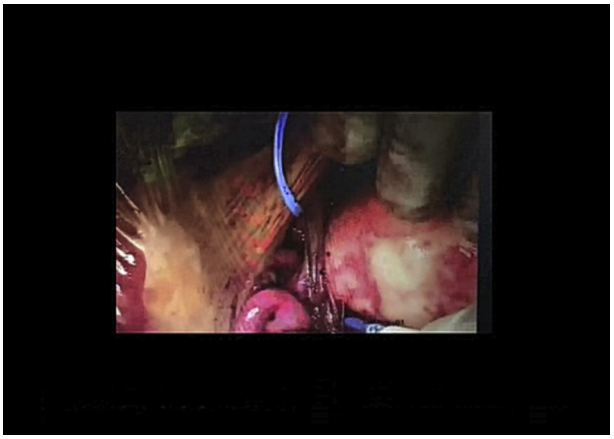



**FIGURE 1.** A, Preoperative computed tomographic 3D reconstruction showing dilation of ascending and proximal descending aorta with marked tortuosity of the arch vessels and abdominal aorta. The isthmus is spared. B, Transesophageal echocardiography demonstrating dilated aortic root with moderate to severe aortic regurgitation. C, Surgical en face view of the trileaflet aortic valve in diastole (3D echocardiography reconstruction) demonstrating thickened NCC and central noncoaptation. D, Operative image of elongated and tortuous arch vessel and hand-sewn graft. E, Verhoeff's elastic stain (magnification ×100) showed loss of elastic fibers and increase in fibroblasts in the media of the resected aortic specimen. *LV*, Left ventricle; *AO*, ascending aorta; *RCC*, right coronary cusp; *LCC*, left coronary cusp; *NCC*, noncoronary cusp.

straight graft was used for valve-sparing aortic root reimplantation. The graft size corresponded to the height of the commissure between the left and the noncoronary sinus (22 mm). Symmetric suspension of the commissures and saline testing revealed insufficiency due to prolapse and inadequate coaptation along the thickened noncoronary cusp: this was addressed with leaflet thinning and augmentation with a pericardial strip along the free edge of the cusp.

At 18 °C, selective antegrade cerebral perfusion was established through the right carotid artery. The ascending aorta, arch, and proximal descending aorta was replaced en bloc with a hand-sewn 16-mm Dacron graft having 4 branch grafts of 7 mm for head vessels. Cerebral near-infrared spectroscopy was used throughout. The distal anastomosis was comfortable through median sternotomy. Free ends of the 16-mm and 22-mm Dacron grafts were anastomosed to each other while rewarming (Video 2). Selective antegrade cerebral perfusion, crossclamp, and cardiopulmonary bypass times were 40, 207, and 275 minutes, respectively.

Postprocedure echocardiography showed good coaptation of the aortic leaflets with trivial central regurgitation (Video 3). There was no gradient across the neoarch graft to the descending aorta. CTA demonstrated sufficient graft design. Histopathology showed loss of elastic fibers in the aortic media (Figure 2). Annual surveillance echocardiogram demonstrated a competent aortic valve with trivial

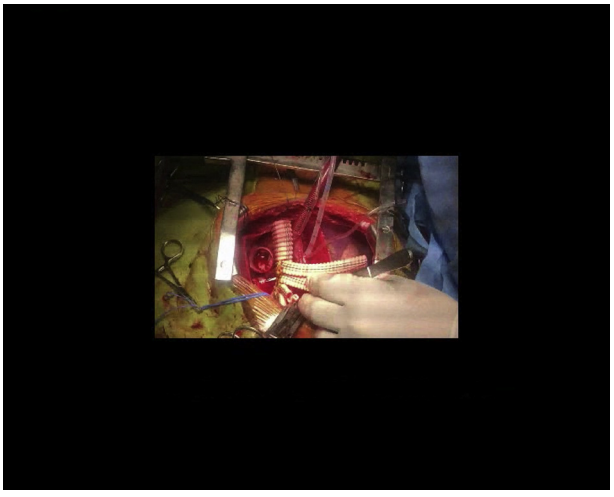


**VIDEO 1.** Operative view of cutis laxa aortopathy with giant thoracic aortic aneurysm and tortuous arch vessels. Video available at: https://www.jtcvs.org/article/S2666-2507(22)00259-0/fulltext.

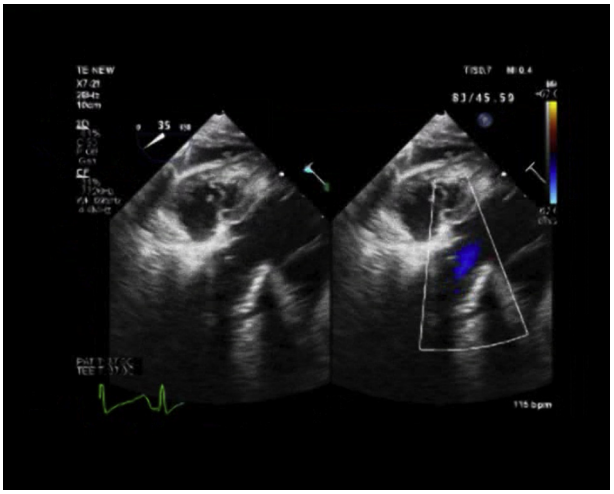
AR. The patient has completed an uneventful 18-month follow-up. Life-long surveillance of the entire arterial tree is recommended.[3]

## COMMENT

ARCL 1B is characterized by loss of function mutations in the EFEMP2 (alias FBLN4) gene, which encodes the extracellular matrix protein fibulin-4.[3] Failure of cross-linking elastin and collagen leads to arterial wall weakness, resulting in aneurysm formation and arterial elongation. The ascending aorta and arch vessels are more affected with distinctive sparing of the aortic isthmus.[3-6] Additional morbidity is related to airway compression (respiratory distress), esophageal compression (feeding difficulty), and stenosis of cerebral vessels (seizures).



**VIDEO 2.** Custom-made prosthetic graft replacement of diseased aortic segments. Video available at: https://www.jtcvs.org/article/S2666-2507(22)00259-0/fulltext.



**VIDEO 3.** Echocardiography showing pre- and postrepair aortic regurgitation. Video available at: https://www.jtcvs.org/article/S2666-2507(22)00259-0/fulltext.

No clinical practice guidelines for EFEMP2-related cutis laxa have been published.[3] Kappanayil and colleagues[4] reported 80% mortality at a median age of 4 months. There are only 2 reports to date on surgical intervention in cutis laxa for aneurysmal involvement beyond the ascending aorta. Hebson and colleagues[5] described a 2-stage elephant trunk technique to address combined ascending and descending aortic aneurysms (intersurgical interval of 9 months) without aortic valve intervention. Yetman and colleagues[6] reported valve reimplantation along with aortic root and total arch replacement. Arterial tortuosity was not noted. Our patient had a large root with advanced AR. We chose a valve-sparing replacement over a Bentall procedure to avoid the need for long-term anticoagulation in young girl. To the best of our knowledge, this is the first description of intervention in cutis laxa aortopathy with aortic leaflet pathology and extensive thoracic aortic involvement from aortic root to descending aorta, successfully addressed in a single setting. A multidisciplinary approach played a key role in defining pathology and decision-making.

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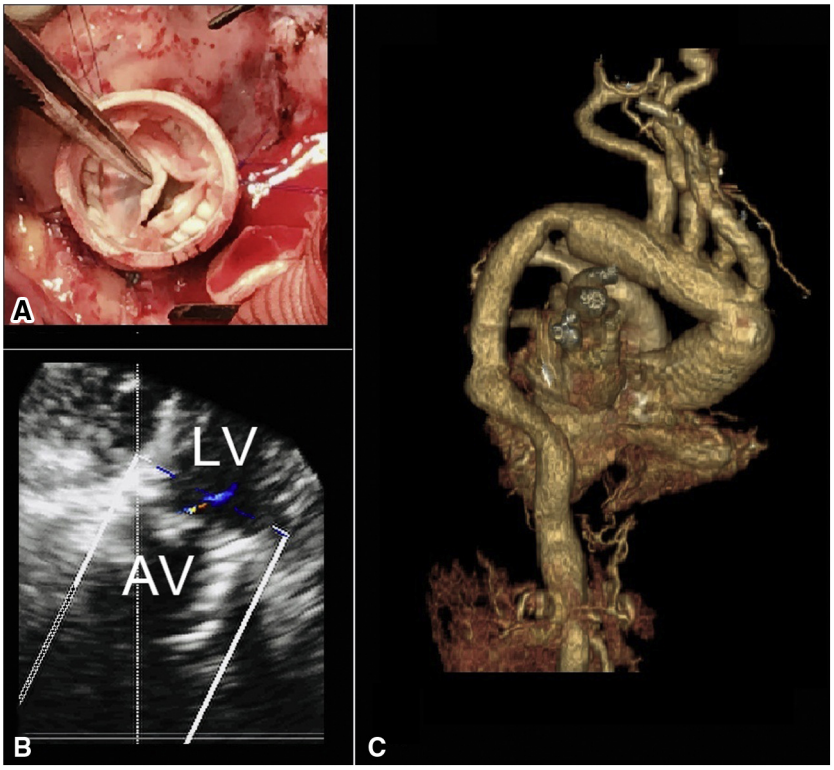


**FIGURE 2.** A, Intraoperative view of the reimplanted trileaflet aortic valve. B, Postprocedure echocardiogram demonstrating good leaflet coaptation and trace regurgitation. C, Postoperative 3-dimensional computed tomographic reconstruction of the composite graft. *LV*, Left ventricle; *AV*, aortic valve.


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